



Variation Resources

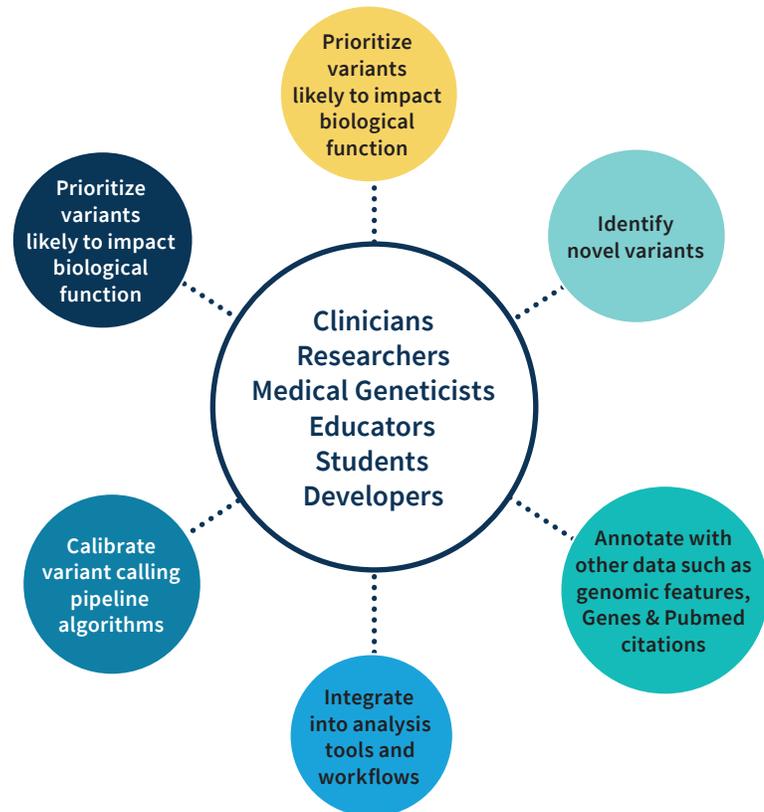
NCBI's variation resources offer human genomic variations including both common and rare SNV, other small scale variations, and large structural variations. Access this information for research and/or clinical interpretation using our websites, FTP, and API services.

dbSNP

- 695 Million RS
- Frequency for more than 552 Million RS; including common and rare variants
- Rich annotation reported on RefSeq GRCH37 and GRCH38 assemblies, mRNA, and Protein

dbVar

- 174 studies
- Clinically significant SV, Case-Control, and Curated Datasets
- 4.2 million unique structural variants
- 35.9 million submitted variant calls
- Updated monthly



Learn More About Our Variation Resources

dbSNP

- VCF files for assemblies GRCh37 and GRCh38
- Full set of RefSNPs in the JSON format
- <https://go.usa.gov/xVPdD>

Variation Services

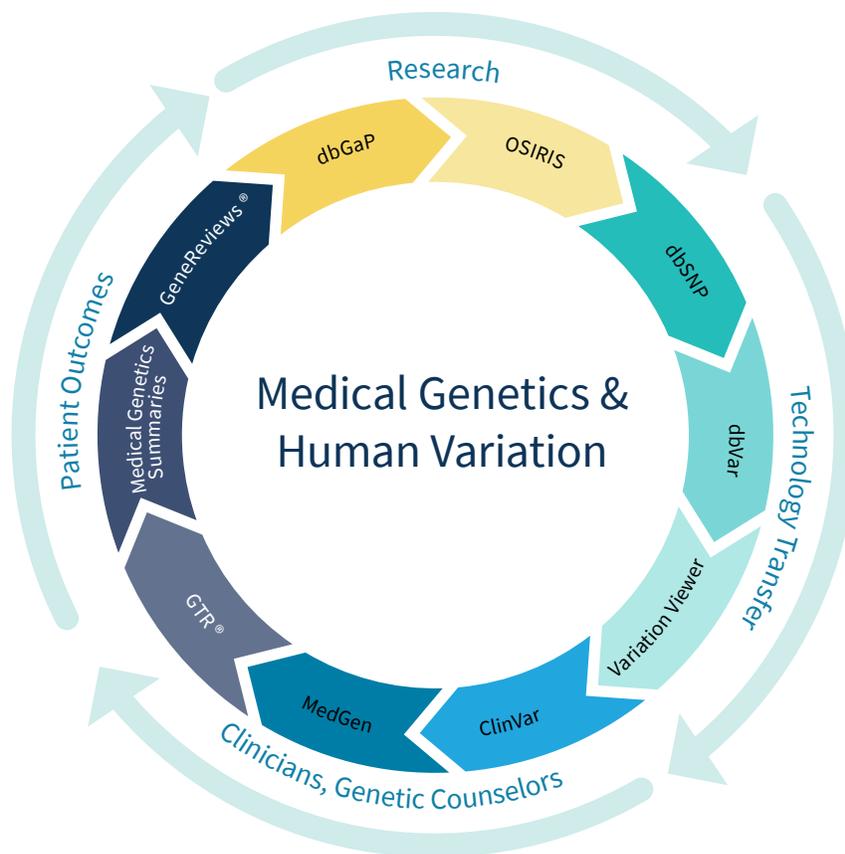
- Web services for comparing, normalizing, annotating, and interconverting (take out grouping)
- <https://go.usa.gov/xVPdW>

dbVar

- Files are available in XML, GVF, VCF, BED, BEDPE, and TSV for assemblies GRCh37 and GRCh38
- <http://bit.ly/2koP2Zg>
- Full set of FTP files described in <https://go.usa.gov/xVPdZ>

Variation Viewer

- View, search, and navigate variations in genomic context. Review data from dbSNP, dbVar and ClinVar, or upload your own data
- <https://go.usa.gov/xVPdK>



ClinVar

user-submitted database for information about genomic variation and its relationship to human health.



dbGaP

database for genotype and phenotype research studies.



dbSNP and dbVar

databases of small and large genomic variants including both common variations and clinical mutations.



GTR®

provider-submitted database of clinical and research molecular, cytogenetic and biochemical genetic tests and supporting information.



MedGen

aggregates information from and provides access to authoritative medical genetics resources.



Medical Genetics Summaries and GeneReviews®

up-to-date, peer-reviewed, medically actionable summaries for heritable diseases and pharmacogenetics.



OSIRIS

Open source short tandem repeat (STR) analysis tool for forensic, clinical and research use.



Variation Viewer

interactive browser for examination of nucleotide variants in a genomic context.